

## Association of bilateral, multiple presumed retinal astrocytic proliferations with combined hamartoma of retina and retinal pigment epithelium in a 9-year-old male child with neurofibromatosis type 2

Pukhraj Rishi, Raj Shri Hirawat, Aditya Verma

Neurofibromatosis type 2 (NF-2) is characterized by multifocal proliferation of neural crest-derived cells. The characteristic finding of NF-2 is bilateral vestibular schwannomas. Combined hamartoma of retina and retinal epithelium (CHRRPE) is another associated finding. A 9 year-old-male child presented with left eye decreased vision for 3 months. Visual acuity was 0.0 and 0.8 LogMAR in the right and left eye, respectively. Left fundus showed an elevated, pigmented lesion with surface wrinkling and vascular tortuosity suggestive of CHRRPE with multiple presumed retinal astrocytic proliferations in mid-periphery. He had multiple café-au-lait spots. Optical coherence tomography confirmed clinical findings. Magnetic resonance imaging brain showed bilateral acoustic neuroma. Recognition of this rare finding as presenting feature of NF-2 can lead to earlier diagnosis which is vital to appropriate surveillance and possible surgical intervention. It is recommended that children with CHRRPE be screened for NF-2.

**Key words:** Combined hamartoma of retina and retinal pigment epithelium, eye tumor, neurofibromatosis type 2, retinal astrocytic proliferation

To report an unusual case of combined hamartoma of retina and retinal pigment epithelium (CHRRPE), bilateral multiple presumed retinal astrocytic proliferations (MPRAP) in a 9-year-old male child with neurofibromatosis type 2 (NF-2).<sup>[1-3]</sup> NF-2 is characterized by multifocal proliferation of neural crest-derived cells. The characteristic finding of NF-2 is a bilateral vestibular schwannoma. CHRRPE is another associated finding. Association of bilateral MPRAP with NF-2 has not been described before.

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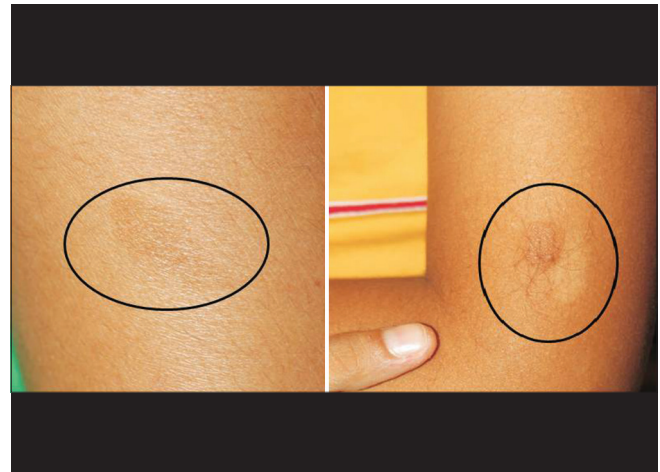
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## Case Report

A 9-year-old male child presented with decreased vision in the left eye of 3 months duration. Visual acuity was 0.0 and 0.8 LogMAR in the right and left eye, respectively. General examination revealed multiple café-au-lait skin spots over the back and arms [Fig. 1]. Fundus examination revealed bilateral MPRAP and CHRRPE in the left eye [Figs. 2 and 3]. Suspecting NF-2, neuroimaging was done; magnetic resonance imaging revealed bilateral acoustic neuroma [Fig. 4]. Optical coherence tomography (OCT) showed vitreoretinal traction in both eyes and disorganized inner retinal structures in the left eye [Fig. 5]. Fundus fluorescein angiography of MPRAP showed central hypofluorescence (blocked fluorescence) with surrounding hyperfluorescence (staining) [Fig. 6].

## Discussion

Presumed solitary circumscribed retinal astrocytic proliferation is a recently described entity that occurs in middle-aged to older patients.<sup>[4]</sup> It appears as a small, solitary, well-defined opaque lesion confined to the retina. It is not associated with subretinal fluid, exudation, or feeder vessel. Our case exhibits the defined clinical characteristics but manifests with multiple lesions bilaterally in a 9-year-old boy, associated with CHRRPE that we describe as MPRAP. OCT scan through the presumed retinal astrocytic proliferation in the right eye showed characteristics of retinal astrocytic hamartoma (RAH) type II, as described by Pichi *et al.*<sup>[5]</sup> Neuroimaging revealed bilateral acoustic neuroma suggestive of NF-2. Hence, it is possible to have multiple retinal astrocytic proliferations in a case of NF-2.

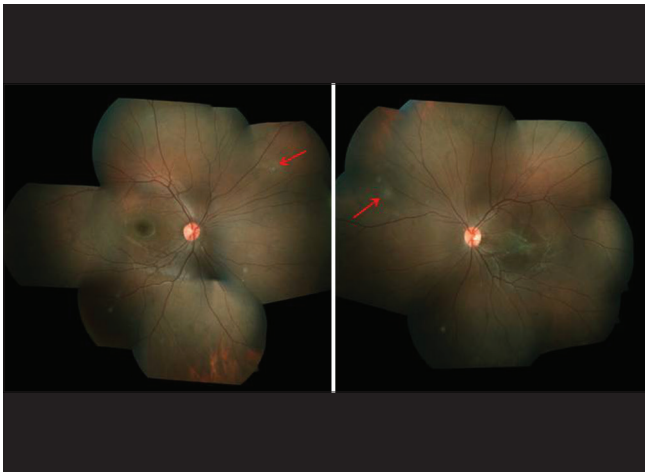


**Figure 1:** Systemic examination revealed multiple café-au-lait spots over skin

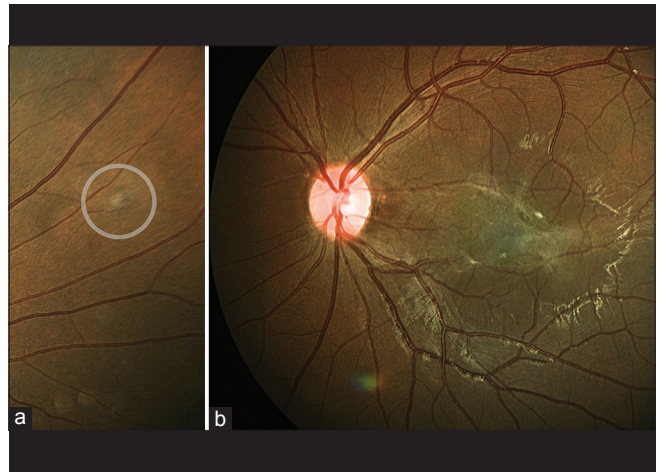
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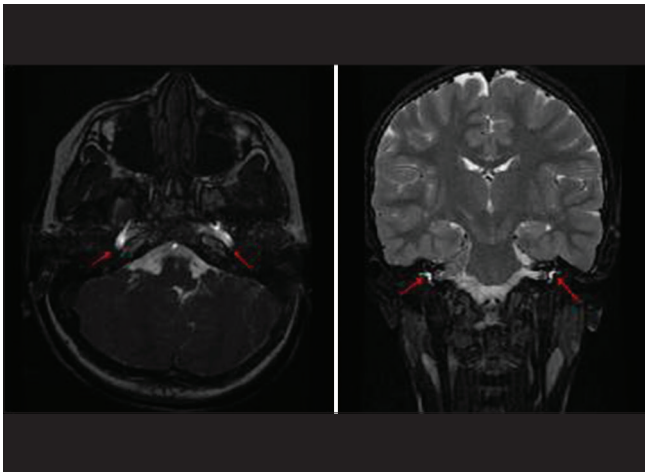
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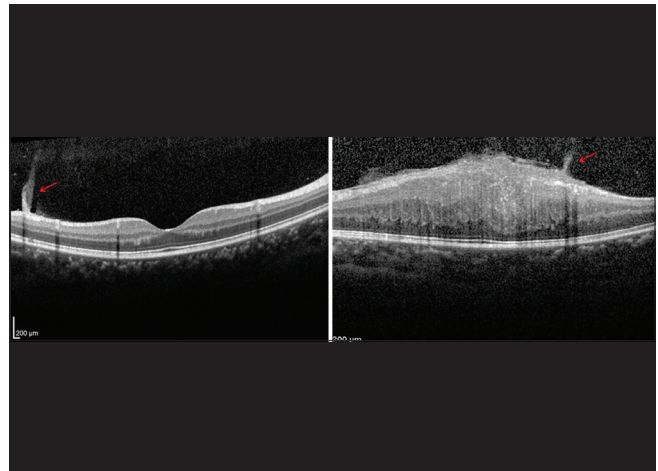
**Figure 2:** Left eye fundus showed elevated, pigmented macular lesion with surface wrinkling and vascular tortuosity suggestive of combined hamartoma of retina and retinal epithelium. Both eye fundi showed multiple presumed retinal astrocytic proliferations (arrows)



**Figure 3:** Presumed retinal astrocytic proliferation (circle) in superonasal quadrant of right eye (a), and closer view of combined hamartoma of retina and retinal epithelium in left eye (b)



**Figure 4:** Bilateral acoustic neuroma on magnetic resonance imaging (arrows)



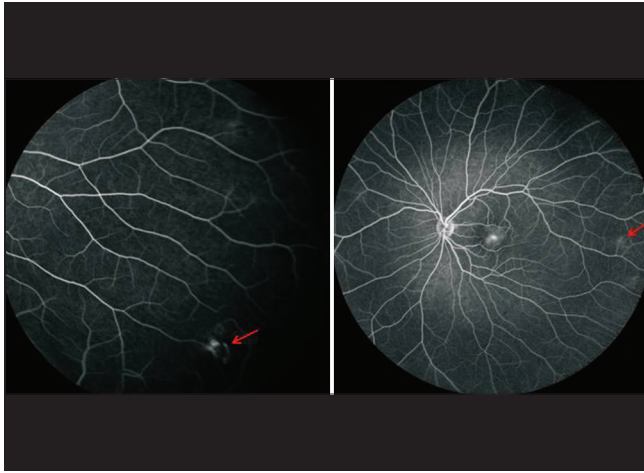
**Figure 5:** Optical coherence tomography showed vitreoretinal traction in both eyes (arrows) and disorganized inner retinal structures in the left eye

The differential diagnosis of a well-circumscribed yellow-white lesion of the retina includes RAH, acquired retinal astrocytoma, retinoblastoma, retinocytoma, myelinated retinal nerve fibers, granuloma, reactive gliosis, unifocal helioid choroiditis (solitary idiopathic choroiditis), and solitary circumscribed retinal astrocytic proliferation.<sup>[6-8]</sup>

RAH is typically diagnosed early in life and often associated with tuberous sclerosis. In such cases, lesions are often multiple or bilateral. Our patient did not have any of the stigmata of tuberous sclerosis but presented with cafe-au-lait spots, CHRRPE, and neuroimaging revealed bilateral acoustic neuroma - all suggestive of NF-2. Acquired retinal astrocytomas tend to be progressive and may be associated with exudation. These lesions may correspond to astrocytomas of central nervous system. In our case, lesions lack exudation. Retinocytomas appear as gray translucent tumors with intralesional calcification, surrounding retinal pigment epithelium alterations, and intralesional cysts.<sup>[8]</sup> Our case had smaller lesions than what is typically seen with

retinocytoma and did not have the characteristic features of that lesion. Unifocal helioid choroiditis (solitary idiopathic choroiditis) also presents with a yellow-white circumscribed lesion in the posterior pole. The active phase may present with overlying neurosensory detachment and occasional vitreous cell. A discrete yellow-white choroidal lesion is hallmark of inactive phase. Our case differs because of its location within retina, rather than choroid.

In summary, our patient has multiple bilateral circumscribed astrocytic proliferations. These lesions are well-circumscribed, opaque intraretinal lesions without exudation or feeding vessels. These differ from other white or yellow-white lesions of the retina in important ways that enable the clinician to appropriately image the patient for possible features of NF-2. Hence, recognition of this rare finding as presenting feature of NF-2 can lead to earlier diagnosis which is vital to appropriate surveillance and possible surgical intervention. It is recommended that children with CHRRPE/MPRAP be screened for NF-2.



**Figure 6:** Fundus fluorescein angiography showed central hypofluorescence (blocked fluorescence) with surrounding hyperfluorescence (staining)

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#### Conflicts of interest

There are no conflicts of interest.

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